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08/509,359

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☐ (A) referred to in:

United States Patent Application Publication No. _____, page _____, line _____,

United States Patent Number 5,986,054, column _____, line _____, or

an International Application which was filed on or after November 29, 2000 and which

designates the United States, WIPO Pub. No. _____, page _____, line _____.

- ☐ (B) referred to in an application that is open to public inspection as set forth in 37 CFR 1.11(b) or 1.14(e)(2)(i), i.e., Application No. _____, paper No. _____, page _____, line _____.

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US005986054A

United States Patent [19]**St. George-Hyslop et al.**[11] **Patent Number:** **5,986,054**[45] **Date of Patent:** **Nov. 16, 1999****[54] GENETIC SEQUENCES AND PROTEINS
RELATED TO ALZHEIMER'S DISEASE****[75] Inventors:** Peter H. St. George-Hyslop; Johanna M. Rommens; Paul E. Fraser, all of Toronto, Canada**[73] Assignees:** The Hospital for Sick Children, HSC Research and Development Limited Partnership; The Governing Council of the University of Toronto, both of Canada**[21] Appl. No.:** 08/592,541**[22] Filed:** Jan. 26, 1996**Related U.S. Application Data****[63]** Continuation-in-part of application No. 08/509,359, Jul. 31, 1995, which is a continuation-in-part of application No. 08/496,841, Jun. 28, 1995, which is a continuation-in-part of application No. 08/431,048, Apr. 28, 1995.**[51] Int. Cl.⁶** C07K 14/00; C12P 21/06**[52] U.S. Cl.** 530/350; 435/69.1**[58] Field of Search** 530/350; 435/69.1**[56] References Cited****U.S. PATENT DOCUMENTS**5,262,332 11/1993 Selkoe 436/518
5,297,562 3/1994 Potter 128/898**FOREIGN PATENT DOCUMENTS**2054302 4/1992 Canada .
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(List continued on next page.)

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The present invention describes the identification, isolation and cloning of two human presenilin genes, PS-1 and PS-2, mutations in which lead to Familial Alzheimer's Disease. Also identified are presenilin homologue genes in mice, *C. elegans* and *D. melanogaster*. Transcripts and products of these genes are useful in detecting and diagnosing Alzheimer's disease, developing therapeutics for treatment of Alzheimer's disease, as well as the isolation and manufacture of the protein and the constructions of transgenic animals expressing the mutant genes.

29 Claims, 12 Drawing Sheets

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